Mayer-Rokitansky-Küster-Hauser syndrome

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a disorder that occurs in females and mainly affects the reproductive system. This condition causes the vagina and uterus to be underdeveloped or absent. Affected women usually do not have menstrual periods due to the absent uterus. Often, the first noticeable sign of MRKH syndrome is that menstruation does not begin by age 16 (primary amenorrhea). Women with MRKH syndrome have a female chromosome pattern (46,XX) and normally functioning ovaries. They also have normal female external genitalia and normal breast and pubic hair development. Although women with this condition are usually unable to carry a pregnancy, they may be able to have children through assisted reproduction.

Women with MRKH syndrome may also have abnormalities in other parts of the body. The kidneys may be abnormally formed or positioned, or one kidney may fail to develop (unilateral renal agenesis). Affected individuals commonly develop skeletal abnormalities, particularly of the spinal bones (vertebrae). Females with MRKH syndrome may also have hearing loss or heart defects.

Frequency

MRKH syndrome affects approximately 1 in 4,500 newborn girls.

Genetic Changes

The cause of MRKH syndrome is unknown, although it probably results from a combination of genetic and environmental factors. Researchers have not identified any genes associated with MRKH syndrome.

The reproductive abnormalities of MRKH syndrome are due to incomplete development of the Müllerian duct. This structure in the embryo develops into the uterus, fallopian tubes, cervix, and the upper part of the vagina. The cause of the abnormal development of the Müllerian duct in affected individuals is unknown. Originally, researchers believed that MRKH syndrome was caused by something the fetus was exposed to during pregnancy, such as a medication or maternal illness. However, studies have not identified an association with maternal drug use, illness, or other factors. It is also unclear why some affected individuals have abnormalities in parts of the body other than the reproductive system.

Inheritance Pattern

Most cases of MRKH syndrome occur in people with no history of the disorder in their family.

Less often, MRKH syndrome is passed through generations in families. Its inheritance pattern is usually unclear because the signs and symptoms of the condition frequently vary among affected individuals in the same family. However, in some families, the condition appears to have an autosomal dominant pattern of inheritance. Autosomal dominant inheritance means that one copy of the altered gene in each cell is typically sufficient to cause the disorder, although no genes have been associated with MRKH syndrome.

Other Names for This Condition

- congenital absence of the uterus and vagina (CAUV)
- genital renal ear syndrome (GRES)
- MRKH syndrome
- Mullerian agenesis
- Mullerian aplasia
- Mullerian dysgenesis
- Rokitansky syndrome

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Rokitansky Kuster Hauser syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1698581/

Other Diagnosis and Management Resources

- Children's Hospital Boston: Center for Young Women's Health
  http://youngwomenshealth.org/2013/10/02/mrkh/

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Developmental Disorders of the Female Reproductive Tract
  https://medlineplus.gov/ency/article/001497.htm
- Encyclopedia: Primary Amenorrhea
  https://medlineplus.gov/ency/article/001218.htm
- Health Topic: Uterine Diseases
  https://medlineplus.gov/uterinediseases.html

Genetic and Rare Diseases Information Center

- Mullerian aplasia
  https://rarediseases.info.nih.gov/diseases/7100/mullerian-aplasia

Additional NIH Resources

- Eunice Kennedy Shriver National Institute of Child Health and Human Development: Amenorrhea
  https://www.nichd.nih.gov/health/topics/amenorrhea/Pages/default.aspx

Educational Resources

- Children's Hospital Boston: Center for Young Women's Health
  http://youngwomenshealth.org/2013/10/02/mrkh/
- Disease InfoSearch: Rokitansky Kuster Hauser syndrome
  http://www.diseaseinfosearch.org/Rokitansky+Kuster+Hauser+syndrome/6355
- Kids Health from the Nemours Foundation
  http://kidshealth.org/en/parents/female-reproductive-system.html
- MalaCards: mayer-rokitansky-kuster-hauser syndrome
  http://www.malacards.org/card/mayer_rokitansky_kuster_hauser_syndrome
- Orphanet: Mayer-Rokitansky-Küster-Hauser syndrome
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=3109

Patient Support and Advocacy Resources

- Accord Alliance
  http://www.accordalliance.org/
- Beautiful You MRKH Foundation
  https://www.beautifulyoumrkh.org/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/mayer-rokitansky-kuster-hauser-syndrome/

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22Mayer-Rokitansky-Kuster-Hauser+syndrome%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28mayer-rokitansky-kuster-hauser+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D

OMIM

- MAYER-ROKITANSKY-KUSTER-HAUSER SYNDROME
  http://omim.org/entry/277000
- MULLERIAN DUCT APLASIA, UNILATERAL RENAL AGENESIS, AND CERVICOTHORACIC SOMITE ANOMALIES
  http://omim.org/entry/601076

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